RMRP gene

RNA component of mitochondrial RNA processing endoribonuclease

Normal Function

Unlike many genes, the *RMRP* gene does not contain instructions for making a protein. Instead, a molecule called a noncoding RNA, a chemical cousin of DNA, is produced from the *RMRP* gene. This RNA attaches (binds) to several proteins, forming an enzyme called mitochondrial RNA-processing endoribonuclease, or RNase MRP.

The RNase MRP enzyme is thought to be involved in several important processes in the cell. For example, it likely helps copy (replicate) the DNA found in the energy-producing centers of cells (mitochondria). The RNase MRP enzyme probably also processes ribosomal RNA, which is required for assembling protein building blocks (amino acids) into functioning proteins. In addition, this enzyme helps control the cell cycle, which is the cell's way of replicating itself in an organized, step-by-step fashion.

Health Conditions Related to Genetic Changes

cartilage-hair hypoplasia

More than 50 mutations that cause cartilage-hair hypoplasia have been identified in the *RMRP* gene. Approximately 90 percent of cases of this disorder result from a mutation in which the DNA building block (nucleotide) guanine is substituted for the nucleotide adenine at position 70 in the *RMRP* gene (written as 70A>G). This mutation is found in almost all known affected individuals from the Amish population, approximately 92 percent of those of Finnish descent, and about half of those in other populations.

Mutations in the *RMRP* gene likely result in the production of a noncoding RNA that is unstable. This unstable molecule cannot bind to some of the proteins needed to make the RNase MRP enzyme complex. These changes are believed to affect the activity of the enzyme, which interferes with its important functions within cells. Disruption of the RNase MRP enzyme complex causes short stature (dwarfism), skeletal abnormalities, abnormal immune system function (immune deficiency), elevated cancer risk, sparse hair growth (hypotrichosis), and other signs and symptoms of cartilage-hair hypoplasia.

other disorders

Mutations in the *RMRP* gene can cause other disorders that, like cartilage-hair hypoplasia, result in malformations near the ends of long bones in the arms and legs (metaphyseal dysplasia). *RMRP* gene mutations, including some of the same

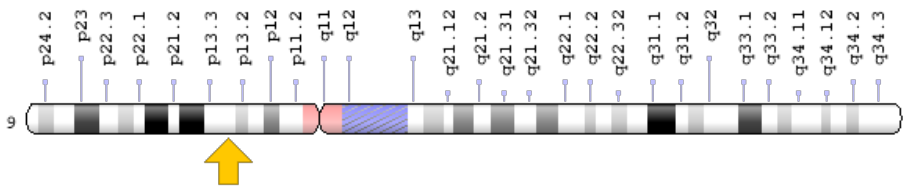
mutations that cause cartilage-hair hypoplasia, may result in a similar disorder known as metaphyseal dysplasia without hypotrichosis. This condition is characterized by short stature and skeletal abnormalities that are usually less pronounced than those seen in cartilage-hair hypoplasia. Individuals with metaphyseal dysplasia without hypotrichosis do not have any changes in the structure or appearance of their hair, but they may have immune deficiency and an increased risk of developing cancer. It is unknown why the same mutations may cause both these conditions.

Anauxetic dysplasia is caused by different *RMRP* mutations than those that cause cartilage-hair hypoplasia and metaphyseal dysplasia without hypotrichosis. People with anauxetic dysplasia have extreme short stature and severe skeletal abnormalities. This condition generally does not affect the immune system or the hair, but mild intellectual disability has been reported.

Chromosomal Location

Cytogenetic Location: 9p13.3, which is the short (p) arm of chromosome 9 at position 13.3

Molecular Location: base pairs 35,657,751 to 35,658,018 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CHH
- NME1
- RMRPR
- RRP2

Additional Information & Resources

GeneReviews

- Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders https://www.ncbi.nlm.nih.gov/books/NBK84550

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28RMRP%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ANAUXETIC DYSPLASIA
  http://omim.org/entry/607095
- CARTILAGE-HAIR HYPOPLASIA
  http://omim.org/entry/250250
- METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS
  http://omim.org/entry/250460
- MITOCHONDRIAL RNA-PROCESSING ENDORIBONUCLEASE, RNA COMPONENT OF
  http://omim.org/entry/157660

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/RMRPID44001ch9p21.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RMRP%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10031
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6023

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/RMRP

Reviewed: November 2008
Published: March 21, 2017

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